

Claim Listing

Claim 1. (currently amended) A method for interrogating genetic variations comprising:

obtaining a plurality of functional regions of the genome, wherein the functional regions comprise at least 10,000 bases;

obtaining information on the functional regions of the genome, using method selection from a group consisting of transcription sites, binding site, origin of replication, and chromatin modification sites;

performing a sequence variation detection assay wherein the information is used to design the assay; and

determining sequence variations of a plurality of individuals in the functional regions of the genome.

Claim 2. (original) The method of claim 1 wherein the functional regions comprise a plurality of transcription factor binding sites.

Claim 3. (withdrawn) The method of claim 2 wherein the functional regions comprise a plurality of RNA:protein binding domains.

Claim 4. (withdrawn) The method of claim 3 wherein the functional regions comprise a plurality of chromatin modification sites.

Claim 5. (withdrawn) The method of claim 4 wherein the functional regions comprise a plurality of origins of replication.

Claim 6. (withdrawn) The method of claim 5 wherein the functional regions comprise a plurality of DNA methylation sites.

Claim 7. (original) The method of claim 1 wherein the obtaining comprises determining functional regions using microarrays.

Claim 8. (original) The method of claim 7 wherein the microarrays are high density oligonucleotide arrays.

Claim 9. (original) The method of claim 8 wherein the microarrays comprise oligonucleotide probes tiling regions of the genome.

Claim 10. (original) The method of claim 9 wherein the determining comprises determining the sequences of the functional regions of a plurality of individuals.

Claim 11. (original) The method of claim 9 wherein the determining comprises determining the genotypes of the functional regions of a plurality of individuals.

Claim 12. (original) The method of claim 11 wherein the genotypes are SNP genotypes.

Claim 13. (original) The method of claim 12 wherein the determining comprises performing a WGSa with at least one restriction enzyme that is suitable for interrogating at least one functional region.

Claim 14. (original) The method of claim 9 wherein the determining comprises determining sequence copy number changes.

Claim 15. (original) The method of claim 1 wherein the functional regions comprise at least 100000 bases.

Claim 16. (original) The method of claim 15 wherein the functional regions comprise at least 500000 bases.

Claim 17. (currently amended) A method for interrogating genetic variations comprising:

obtaining at least one interested genomic segment;

obtaining a plurality of functional regions within the interested genomic segment,
wherein the functional regions comprise at least 5,000 bases;

obtaining information on the functional regions of the genome, using method
selection from a group consisting of transcription sites, binding site, origin of replication,
and chromatin modification sites;

performing a sequence variation detection assay wherein the information is used
to design the assay; and

determining sequence variations of a plurality of individuals in the functional
regions of the genome.

Claim 18. (original) The method of claim 17 wherein the interested genomic
region is determined by association or linkage analysis.

Claim 19. (original) The method of claim 18 wherein the functional regions
comprise a plurality of transcription factor binding sites.

Claim 20. (withdrawn) The method of claim 18 wherein the functional regions
comprise a plurality of RNA:protein binding domains.

Claim 21. (withdrawn) The method of claim 18 wherein the functional regions
comprise a plurality of chromatin modification sites.

Claim 22. (withdrawn) The method of claim 18 wherein the functional regions
comprise a plurality of origins of replication.

Claim 23. (withdrawn) The method of claim 18 wherein the functional regions
comprise a plurality of DNA methylation sites.

Claim 24. (original) The method of claim 17 wherein the obtaining comprises determining functional regions using microarrays.

Claim 25. (original) The method of claim 24 wherein the microarrays are high density oligonucleotide arrays.

Claim 26. (original) The method of claim 25 wherein the microarrays comprise oligonucleotide probes tiling regions of the genome.

Claim 27. (original) The method of claim 24 wherein the determining comprises determining the sequences of the functional regions of a plurality of individuals.

Claim 28. (original) The method of claim 24 wherein the determining comprises determining the genotypes of the functional regions of a plurality of individuals.

Claim 29. (original) The method of claim 28 wherein the genotypes are SNP genotypes.

Claim 30. (original) The method of claim 29 wherein the determining comprises performing a WGS with at least one restriction enzyme that is suitable for interrogating at least one functional region.

Claim 31. (original) The method of claim 24 wherein the determining comprises determining sequence copy number changes.